Statutory Approvals Committee - minutes

Centre 0327 (Boston Place)

Preimplantation Genetic Diagnosis (PGD) application for Gastrointestinal defects and immunodeficiency syndrome, OMIM #243150

Date:	25 February 2021
Venue:	Microsoft Teams Meeting
Committee Members:	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde
Specialist Adviser:	Alan Fryer
Legal Adviser:	Sarah Ellson - FieldFisher
Members of the Executive:	Moya Berry - Committee officer Catherine Burwood - Licensing Manager
Observers:	Sarah Steadman - Inspector (Induction) Karen Campbell - Inspector (Induction)
Apologies:	No apologies were received for the meeting
Declarations of Interest:	Members of the committee declared that they had no conflicts of interest in relation to this item.

The Committee had before it:

- HFEA Code of Practice 9th edition
- Standard Licencing and Approvals Pack

The following papers were considered by the committee:

- Executive Summary
- PGD Application form
- Redacted Peer review

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr Alan Fryer, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Gastrointestinal defects and immunodeficiency syndrome (GIDID), OMIM #243150 is consistent with the peer review.

1.3. The committee noted that the condition being applied for is not on the list of approved PGD conditions.

1.4. The committee noted that a Genetic Alliance (UK) statement had not been provided for this application.

1.5. The committee had regard to its decision tree. The committee noted that the Centre is licensed to carry out PGD. The committee was also satisfied that the Centre has experience of carrying out PGD and that generic patient information about its PGD programme and associated consent forms had previously been received by the HFEA.

1.6. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of Schedule 2 of the Act, i.e., 'where there is a particular risk that the embryo may have any gene, chromosome or mitochondrion abnormality, establishing whether it has that abnormality or any other gene, chromosome or mitochondrion abnormality'.

1.7. The committee noted that Gastrointestinal defects and immunodeficiency syndrome (GIDID), OMIM #243150, is inherited in an autosomal recessive manner, which means there is a 25% chance of an embryo being affected by the condition in each pregnancy if each parent has a relevant mutation.

1.8. The committee noted that the penetrance of the condition is 100%.

1.9. Gastrointestinal defects and immunodeficiency syndrome (GIDID), OMIM #243150 is characterised by multiple intestinal atresia (the abnormal closure or absence of an orifice or passage in the body, in this condition the intestine). The regions of atretic bowel occur at various levels throughout the small and large intestines. Some patients exhibit inflammatory bowel disease (IBD), with or without intestinal atresia, and in some cases, the intestinal features can be associated with mild to severe combined immunodeficiency resulting in recurrent episodes of sepsis. This disorder results in death before the age of 2 years in 70% of patients. Patients classically present with intestinal obstruction prenatally or at birth and the complications, including sepsis, often lead to multiple organ failure.

1.10. There is no cure for this condition and surgical interventions are mostly palliative and have poor outcomes. Surgical procedures (often multiple) involve resection of multiple atretetic bowel segments that may lead to a short bowel syndrome and a requirement for total parenteral nutrition with its associated risks of complications including liver failure. Some patients have then required liver-small bowel transplantation for liver and intestinal failure and bone marrow transplantation for the immunodeficiency.

1.11. The committee noted the executive's request to consider Gastrointestinal defects and immunodeficiency syndrome (GIDID), OMIM #243150, for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that, in the worst-case scenario, Gastrointestinal defects and immunodeficiency syndrome (GIDID), OMIM #243150, is a severe, life-limiting condition that presents prenatally or at birth. The committee noted the peer reviewer's comments that this is a very serious condition that is incompatible with long-term survival. The condition has no effective treatment, and is usually fatal within the first two years of life. The committee considered the devastating impact on those affected with the condition.

2.2. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk that a person with such an abnormality will, given the condition's worst symptoms, have or develop a serious physical disability, a serious illness, or any other serious medical condition.

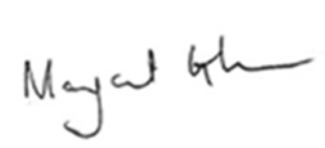
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Gastrointestinal defects and immunodeficiency syndrome (GIDID), OMIM #243150

3. Chair's signature

3.1. I confirm this is a true and accurate record of the meeting,

Signature



Name

Margaret Gilmore

Date

17 March 2021